

# EMMA™ TECHNOLOGY

## METHOD

### High-throughput simultaneous detection of point mutations and large-scale rearrangements by CE

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CE / Heteroduplex analysis / Large-scale rearrangements / Mutation detection

**Abstract** The detection of unknown mutations is important both in population genetics research and in diagnosis. At present, two different methods must be used to detect either point mutations or large-scale genetic rearrangements, which is costly and time-consuming. We describe here a new method for the simultaneous detection of these two types of mutations. It is based on electrophoretic heteroduplex analysis (HDA) using enhanced mismatch mutation analysis (EMMA) and semiquantitative multiplexed PCR conditions.

The use of such conditions allows the simultaneous search of any kind of mutation in up to five different fragments *per* capillary, in a single or multi-CE system. The method was validated on patient samples with mutations in the breast predisposition gene *BRCA1*. It leads to highly reliable and high-throughput mutation detection at low cost, as compared with classical methods.